



## FAM111B gene

family with sequence similarity 111 member B

### Normal Function

The *FAM111B* gene provides instructions for making a protein whose function is not well understood. The FAM111B protein, which is found in many parts of the body, contains a functional region called a peptidase domain. Similar proteins containing such a domain are able to break down other proteins. However, the types of proteins the FAM111B protein interacts with and the roles it plays in the body are unknown.

### Health Conditions Related to Genetic Changes

hereditary fibrosing poikiloderma with tendon contractures, myopathy, and pulmonary fibrosis

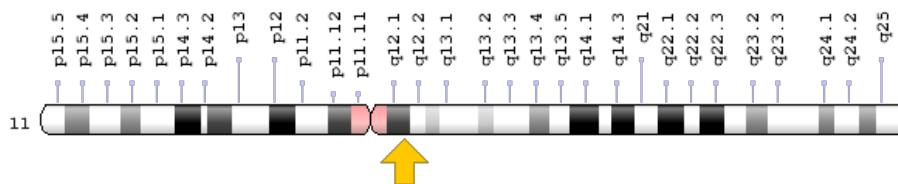
At least six mutations in the *FAM111B* gene have been identified in people with hereditary fibrosing poikiloderma with tendon contractures, myopathy, and pulmonary fibrosis (POIKTMP). This disorder affects many parts of the body, particularly the skin, muscles, lungs, and pancreas.

The *FAM111B* gene mutations that cause POIKTMP result in production of an abnormal FAM111B protein from one copy of the gene in each cell. Because most of the *FAM111B* mutations identified in people with POIKTMP change single protein building blocks (amino acids) in the peptidase domain, researchers think that the mutations alter the protein's function, and that these changes in FAM111B function underlie the varied signs and symptoms of POIKTMP.

## Chromosomal Location

Cytogenetic Location: 11q12.1, which is the long (q) arm of chromosome 11 at position 12.1

Molecular Location: base pairs 59,107,185 to 59,127,415 on chromosome 11 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- cancer-associated nucleoprotein
- CANP
- POIKTMP
- protein FAM111B isoform a
- protein FAM111B isoform b

## Additional Information & Resources

### Educational Resources

- Molecular Cell Biology (fourth edition, 2000): Cells Degrade Proteins by Several Pathways  
<https://www.ncbi.nlm.nih.gov/books/NBK21750/#A559>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28FAM111B%5BTIAB%5D%29+OR+%28family+with+sequence+similarity+111+member+B%5BTIAB%5D%29%29+OR+%28CANP%5BTIAB%5D%29+OR+%28POIKTMP%5BTIAB%5D%29+OR+%28cancer-associated+nucleoprotein%5BTIAB%5D%29+OR+%28protein+FAM111B+isoform+a%5BTIAB%5D%29+OR+%28protein+FAM111B+isoform+b%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

## OMIM

- FAMILY WITH SEQUENCE SIMILARITY 111, MEMBER B  
<http://omim.org/entry/615584>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_FAM111B.html](http://atlasgeneticsoncology.org/Genes/GC_FAM111B.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=FAM111B%5Bgene%5D>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=24200](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=24200)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/374393>
- UniProt  
<http://www.uniprot.org/uniprot/Q6SJ93>

## **Sources for This Summary**

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